J Inherit Metab Dis (2017) 40:307
DOI 10.1007/s10545-017-0019-5

BOOK REVIEW

Nenad Blau (ed.). Phenylketonuria and BH4 deficiencies

3rd Edition 2016; 112 pp; 39.80 Euro; ISBN 978-3-8374-1526-1

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Published online: 6 February 2017

This compact book, part of the *UNI-MED* Science series, is a very comprehensive resource for any clinician (including allied health professionals) who is managing patients with phenylketonuria (PKU) or BH4 deficiencies. Several authors of arguably top reputation in this area of Metabolic Medicine contributed to the 11 chapters of the book, which was edited by Prof. Nenad Blau, a world authority in PKU and BH4 deficiencies.

The book chapters are organised in several sections. The first section includes an historical account on PKU, its 'discovery', newborn screening and treatment, as well as pathophysiological aspects of the phenylalanine hydroxylating system. The second section canvases clinical manifestations of this group of disorders, which obviously include mainly neurocognitive or neurological deficits. In that respect, a short description of patients born before the newborn-screening era, or missed in newborn-screening could be of interest. The third section deals with the diagnosis and the genetics of the disorders. The forth section details treatment modalities of this group of disorders, naturally focussing on dietary management. Of particular interest in a short chapter on the collaborative work between the health professional and the patient and family, which is so pivotal for the success of treatment. This section also includes an up-to-date summary of novel treatments of PKU and treatment of BH4 deficiencies. Overall, the text in each chapter is concise, clear and easy to follow. It is supported by short summaries of key points at the beginning of each chapter, and by many tables and figures, which are very helpful. A note to the publisher: some of the tables and figures are difficult to read because of a very small font.

The practicality of this book is further strengthened by the inclusion of Sect. 5 (Annex; note: not Sect. 4), which provides very helpful resources for patients and families (as stated in its title, but for health care teams as well): information about national PKU societies (which should be updated), on-line information and a list of manufacturers and suppliers of special foods for use in PKU as well as a list of recommended books for further reading.

PKU has been considered a paradigm of a successfully diagnosed and treated metabolic disorder, although many questions and controversies around the optimal management of patients with this disorder still exist. Indeed, treatment protocols differ between metabolic centres, and the evidence underlying these protocols is far from optimal. Some of these issues are mentioned, but in the next edition of the book, consideration should be given to the inclusion of a short chapter at the beginning of Section 4 ("Management"), focussing on these controversies, the lack of sufficient, good quality evidence for particular treatment modalities and the difficulties in obtaining such evidence. This could be of great benefit for those who wonder why there are different guidelines and varying protocols in different centres. For example, Sub-sect. 5 in Chapt. 3 ("Rationale for lifelong treatment of PKU to minimise neurological damage"), which seems a bit out of context in its current location, could be transferred to such chapter and be expanded to include data about adult patients (rather than up to age 10 years) in the argument for lifelong treatment.

In summary, this is an excellent addition to any Metabolic Library in any metabolic centre, and will be of great help to those managing patients with disorders of phenylalanine hydroxylation. This book is definitely worth much more than its actual cost in Euro, which is very affordable.

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